



General

Guideline Title

NSGC practice guideline: risk assessment and genetic counseling for hereditary breast and ovarian cancer.

Bibliographic Source(s)

Berliner JL, Fay AM, Cummings SA, Burnett B, Tillmanns T. NSGC practice guideline: risk assessment and genetic counseling for hereditary breast and ovarian cancer. J Genet Couns. 2013 Apr;22(2):155-63. [56 references] [PubMed](#)

Guideline Status

This is the current release of the guideline.

This guideline updates a previous version: Berliner JL, Fay AM, Familial Cancer Risk Special Interest Group of the National Society of Genetic Counselors. Risk assessment and genetic counseling for hereditary breast and ovarian cancer: recommendations of the National Society of Genetic Counselors. J Genet Counsel 2007 Jun;16(3):241-60. [110 references]

Recommendations

Major Recommendations

Recommendation 1

Published guidelines should be consulted regarding the appropriateness of genetic testing, and a more tailored approach to medical management should be addressed.

Recommendation 2

Evaluation of families for hereditary breast and ovarian cancer (HBOC) syndrome should include consideration of other hereditary syndromes in which breast and ovarian cancers are component cancers.

Recommendation 3

Standards of care for accurate risk assessment should be followed when identifying individuals at risk for breast and/or ovarian cancers. Consider established models and published data to estimate the individual's risk of developing cancer, as this will assist in determining appropriate management as summarized in Recommendation 4 (below).

Recommendation 4

Clients should be given management recommendations based on clinical judgment, complemented by risk assessment, family history, and genetic

test results (when appropriate).

Recommendation 5

The genetics consultation should include the provision of extensive client resources including scientific information, psychosocial support, and advocacy.

Recommendation 6

Clients should be engaged in a discussion of the ethical, legal, and social implications of genetic information and testing.

Conclusions

The process of cancer risk assessment and genetic counseling for HBOC syndrome requires many steps, including:

1. Gathering personal medical and family history data
2. Psychosocial assessment
3. Discussion of cancer and mutation risk and how personalized risk estimates are derived
4. Facilitation of the informed consent process through discussion of the risks, benefits, limitations, and likelihood of identifying a mutation with genetic susceptibility testing
5. Results disclosure (if applicable)
6. Discussion of medical management options
7. Review of issues related to genetic discrimination

Genetics professionals are uniquely suited to facilitating clients' understanding of the genetics of cancer, personalized risk calculations, and the potential psychological, social, and medical implications associated with cancer risk assessment and genetic testing. Genetics professionals are also adept at identifying clients who may need additional support, and providing a referral to appropriate mental health care professionals. The above guidelines provide a best practices approach to offering a more uniform manner of delivering comprehensive care for families and individuals facing HBOC syndrome.

Clinical Algorithm(s)

A genetic testing algorithm is available from the [Journal of Genetic Counseling Web site](#) .

Scope

Disease/Condition(s)

Hereditary breast and ovarian cancer

Guideline Category

Counseling

Evaluation

Management

Risk Assessment

Clinical Specialty

Family Practice

Internal Medicine

Medical Genetics

Obstetrics and Gynecology

Oncology

Psychology

Intended Users

Advanced Practice Nurses

Allied Health Personnel

Health Care Providers

Nurses

Physician Assistants

Physicians

Psychologists/Non-physician Behavioral Health Clinicians

Public Health Departments

Social Workers

Guideline Objective(s)

To present a current and comprehensive set of practice recommendations for effective genetic cancer risk assessment, counseling and testing for hereditary breast and ovarian cancer

Target Population

Individuals with, or at increased risk of, hereditary breast and/or ovarian cancer

Interventions and Practices Considered

1. Gathering personal medical and family history data
2. Psychosocial assessment
3. Discussion of cancer and mutation risk and how personalized risk estimates are derived
4. Facilitation of the informed consent process through discussion of the risks, benefits, limitations, and likelihood of identifying a mutation with genetic susceptibility testing
5. Results disclosure (if applicable)
6. Discussion of medical management options
7. Review of issues related to genetic discrimination

Major Outcomes Considered

Not stated

Methodology

Methods Used to Collect/Select the Evidence

Searches of Electronic Databases

Description of Methods Used to Collect/Select the Evidence

The guideline authors searched via Pubmed for published literature from 1990 to July 2012. Reviews and peer-reviewed articles on relevant topics were included. Non-peer-reviewed publications were excluded. The specific search terms used were hereditary breast cancer, genetic counseling, risk assessment models, medical management of HBOC, professional society guidelines, ethical issues for HBOC, genetic testing, BRCA1, BRCA2, hereditary breast cancer syndromes, surveillance, and chemoprevention.

Number of Source Documents

Not stated

Methods Used to Assess the Quality and Strength of the Evidence

Not stated

Rating Scheme for the Strength of the Evidence

Not applicable

Methods Used to Analyze the Evidence

Review

Description of the Methods Used to Analyze the Evidence

Each practice guideline focuses on a clinical or practice-based issue, and is the result of a review and analysis of current professional literature believed to be reliable.

Methods Used to Formulate the Recommendations

Expert Consensus

Description of Methods Used to Formulate the Recommendations

The recommendations were developed by members of the National Society of Genetic Counselors (NSGC).

Rating Scheme for the Strength of the Recommendations

Not applicable

Cost Analysis

A formal cost analysis was not performed and published cost analyses were not reviewed.

Method of Guideline Validation

Not stated

Description of Method of Guideline Validation

Not applicable

Evidence Supporting the Recommendations

Type of Evidence Supporting the Recommendations

The type of supporting evidence is not specifically stated for each recommendation.

Benefits/Harms of Implementing the Guideline Recommendations

Potential Benefits

- Appropriate use of risk assessment, genetic counseling, and testing for hereditary breast and ovarian cancer
- The guidelines provide a best practices approach to offering a more uniform manner of delivering comprehensive care for families and individuals facing hereditary breast and ovarian cancer (HBOC) syndrome

Potential Harms

- Risk assessment for cancer can raise a number of intellectual and psychosocial issues.
 - Clients need to contend with an enhanced understanding of their specific cancer risks, potentially difficult decisions for managing their cancer risks, concerns about discrimination, and worry about possible risks and reactions in their children and other family members.
 - BRCA mutation carriers face some difficult reproductive choices regarding the use of oral contraceptives, tubal ligation, risk-reducing bilateral salpingo-oophorectomy (BSO), pregnancy, and breastfeeding.
- As with all genetic testing, hereditary breast and ovarian cancer (HBOC) syndrome testing has the potential to raise ethical, legal and social issues, both within the family and for society as a whole. Additionally, patients may express concern about the potential for genetic discrimination, particularly with regard to health and life insurance in the event of a positive test result. The fear of discrimination may prevent at-risk individuals from having genetic testing, which in turn could have significant health implications.

Qualifying Statements

Qualifying Statements

- The practice guidelines of the National Society of Genetic Counselors (NSGC) are developed by members of the NSGC to assist genetic counselors and other health care providers in making decisions about appropriate management of genetic concerns, including access to and/or delivery of services. Each practice guideline focuses on a clinical or practice-based issue, and is the result of a review and analysis of current professional literature believed to be reliable. As such, information and recommendations within the NSGC practice guidelines reflect the current scientific and clinical knowledge at the time of publication, are only current as of their publication date, and are subject to change without notice as advances emerge.
- In addition, variations in practice, which take into account the needs of the individual patient and the resources and limitations unique to the institution or type of practice, may warrant approaches, treatments and/or procedures that differ from the recommendations outlined in this guideline. Therefore, these recommendations should not be construed as dictating an exclusive course of management, nor does the use of such recommendations guarantee a particular outcome. Genetic counseling practice guidelines are never intended to displace a health care

provider's best medical judgment based on the clinical circumstances of a particular patient or patient population. Practice guidelines are published by NSGC for educational and informational purposes only, and NSGC does not "approve" or "endorse" any specific methods, practices, or sources of information.

Implementation of the Guideline

Description of Implementation Strategy

An implementation strategy was not provided.

Implementation Tools

Clinical Algorithm

For information about availability, see the *Availability of Companion Documents* and *Patient Resources* fields below.

Institute of Medicine (IOM) National Healthcare Quality Report Categories

IOM Care Need

Living with Illness

Staying Healthy

IOM Domain

Effectiveness

Patient-centeredness

Identifying Information and Availability

Bibliographic Source(s)

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Adaptation

Not applicable: The guideline was not adapted from another source.

Date Released

2007 Jun (revised 2013 Apr)

Guideline Developer(s)

National Society of Genetic Counselors - Medical Specialty Society

Source(s) of Funding

National Society of Genetic Counselors

Guideline Committee

Not stated

Composition of Group That Authored the Guideline

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Financial Disclosures/Conflicts of Interest

Not stated

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Guideline Availability

Electronic copies: Available to subscribers from the [Journal of Genetic Counseling Web site](#) .

Availability of Companion Documents

The following is available:

- Bennett RL, French KS, Resta RG, Doyle DL. Standardized human pedigree nomenclature: update and assessment of the recommendations of the National Society of Genetic Counselors. *J Genet Couns* 2008 Oct;17(5):424-33. Available to subscribers from the [Journal of Genetic Counseling Web site](#) .

Patient Resources

None provided

NGC Status

This NGC summary was completed by ECRI Institute on May 29, 2008. The information was verified by the guideline developer on June 16, 2008. This NGC summary was updated by ECRI Institute on April 19, 2013. The updated information was verified by the guideline developer on

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